Draft New Policy: Do Not Implement

Genetic Testing for Marfan Syndrome, Thoracic Aortic Aneurysms and Dissections, and Related Connective Tissue Disorders

DESCRIPTION

Marfan syndrome (MFS) is a systemic connective tissue disease (CTD) with a high degree of clinical variability and phenotypes overlapping with other syndromes and disorders. The diagnosis of most suspected CTDs can be based on clinical findings and family history. Some of these disorders are associated with a predisposition to the development of progressive thoracic aortic aneurysms and dissection. Accurate diagnosis of one of these syndromes can lead to changes in clinical management, including surveillance of the aorta, and surgical repair of the aorta, when necessary, as well as surveillance for multisystem involvement in syndromic forms of thoracic aortic aneurysms and dissection. Known pathogenic variants are associated with MFS and the other connective tissue disorders that share clinical features with MFS.

The use of genetic testing to establish a diagnosis in an individual with a suspected connective tissue disorder is most useful in individuals who do not meet sufficient clinical diagnostic criteria at the time of initial examination, in individuals who have an atypical phenotype and other connective tissue disorders cannot be ruled out, and in individuals who belong to a family in which a pathogenic variant is known (presymptomatic diagnosis).

POLICY

- Genetic testing for Marfan syndrome, Ehlers-Danlos syndrome type IV, other syndromes associated with thoracic aortic aneurysms and dissections, and related disorders is considered ***medically necessary*** if the medical appropriateness criteria are met. **(See Medical Appropriateness below.)**

- Genetic testing panels for Marfan syndrome, Ehlers-Danlos syndrome type IV, other syndromes associated with thoracic aortic aneurysms and dissections, and other related connective tissue disorders that are not limited to focused genetic testing are considered ***investigational.***

MEDICAL APPROPRIATENESS

- Genetic testing for Marfan syndrome, Ehlers-Danlos syndrome type IV, other syndromes associated with thoracic aortic aneurysms and dissections, and related disorders is considered **medically appropriate** if **ANY ONE** of the following are met:
  - Genetic testing of a symptomatic individual to determine diagnosis if **ALL** of the following are met:
    - Panels comprised entirely of focused genetic testing limited to the following genes: FBN1 and MYH11; ACTA2, TGFBR1 and TGFBR2; and COL3A1
    - Individual has signs and symptoms of a connective tissue disorder
    - A definitive diagnosis cannot be made using established clinical diagnostic criteria (e.g., Ghent criteria, characteristic systemic clinical findings)
  - Targeted familial variant genetic testing for assessing future risk of disease if **ALL** of the following are met:
    - Individual is asymptomatic
    - Known pathogenic variant in the family

IMPORTANT REMINDERS

- Any specific products referenced in this policy are just examples and are intended for illustrative purposes only. It is not intended to be a recommendation of one product over another and is not intended to represent a complete listing of all products available. These examples are contained in the parenthetical e.g., statement.
- We develop Medical Policies to provide guidance to Members and Providers. This Medical Policy relates only to the services or supplies described in it. The existence of a Medical Policy is not an authorization, certification, explanation of benefits or a contract for the service (or supply) that is referenced in the Medical Policy. For a determination of the benefits that a member is entitled to receive under his or her health plan, the Member's health plan must be reviewed. If there is a conflict between the medical policy and a health plan or government program (e.g., TennCare), the express terms of the health plan or government program will govern.

SOURCES

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EFFECTIVE DATE

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